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## LIST OF ACRONYMS

### used in literature on genome research

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Arbitrary primed marker: *AP-PCR*, *DAF*, *DFP*, *ISSR*, *MAAP*, *OP*, *RAMP*, *RAPD*, *SPAR*, *SSR-anchored*,

Non-Arbitrary primed marker: *AFLP*, *ASO*, *ASAP*, *AS-PCR*, *CAPS*, *DAMD*, *DOP-PCR*, *EST*, *ISTR*, *REP-PCR*, *SCAR*, *STMS*, *STR*, *SRFA*, *SSLP*, *SSR*, *SAMPL*, *S-SAP*, *STS*, *VNTR*,

SNP detection systems: *DOL*, *FRET*, *MADGE*, *MASDA*, *MB*, *OLA*, *TaqMan*,

Hybridization based marker: *CFLP*, *GMS*, *RAMCM*, *RFLP*,

Gel systems: *2-DDGE*, *DGGE*, *FIGE*, *PFGE*, *SCGE*, *SSCP*, *TGGE*,

Populations: *DH*, *BIL*, *IRI*, *NIL*, *RIL*,

Molecular methods: *A SSURE B*, *DDRT-PCR*, *HAPPY*, *MATS*, *PRINS*, *RDA*, *REMI*, *REMI-RFLP*, *RICH*, *SDA*,

DNA Amplification methods: *IPCR*, *LCR*, *NASBA*, *PCR*, *RCA*, *SSI*

Miscellaneous: *BAC*, *BSA*, *GS*, *LOD*, *MAS*, *MARS*, *MITE*, *MSAP*, *PIC*, *QTL*, *YAC*

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### AFLP Amplification Fragment Length Polymorphism

synonymous to DAF

Caetano-Anollés G., B.J. Bassam and P.M. Gresshoff (1991) REVIEW DNA amplification fingerprinting: A strategy for genome analysis. *Plant Molec. Biol. Reporter* 9(4):294-307.

### AFLP Amplified Fragment Length Polymorphism

PCR based marker. Not based on arbitrary priming of oligo's, but amplification of specifically selected restriction fragments (see SRFA).

Zabeau M. and P. Vos (1992) *European Patent Application. Publication no. 0 543 858 A1.*

### *AP-PCR* Arbitrary Primed Polymerase Chain Reaction

DNA amplification using a single random primer of 16-20 bases. Products are often analysed on a polyacrylamide gel and detected by autoradiography.

Welsh, J. and M. McClelland (1990) Fingerprinting genomes using PCR with arbitrary primers. *Nucleic Acids Res.* 18:7213-7218.

## ASO Allele Specific Oligo

A special kind of oligo for a PCR reaction. The sequence of the oligo is designed in such a way to allow/inhibit hybridisation at the spot where the mutant (resistant) allele differs from the wildtype (suseptible) allele. (Variant of AS-PCR)

Beckmann J.S. (1988) *REVIEW* Oligonucleotide polymorphisms: A new tool for genomic genetics. *Bio/Technology* 6:161-164.

## ASAP Allele Specific Associated Primers

Similar to ASO or AS-PCR. The sequence of the deca-mer oligo is derived from normal RAPD, which generated an absence/presence polymorphism. These absence/presence polymorphisms do not require electrophoretic separation of the sample. Presence of an amplification product is detected by measuring fluorescence of Ethidium-Bromide stained DNA.

Gu W.K., N.F. Weeden, J. Yu and Wallace DH (1995) Large-scale, cost-effective screening of PCR products in marker-assited selection applications. *Theor. Appl. Genet.* 91:465-470.

## AS-PCR Allele Specific Polymerase Chain Reaction

Refers to amplification of specific alleles, or DNA sequence variants, at the same locus. Specificity is achieved by designing one or both PCR primers so that they partially overlap the site of sequence difference between the amplified alleles (ASO).

Landegren U., R. Kaiser, J. Sanders and L. Hood (1988) DNA diagnostics. Molecular techniques and automation. *Science* 241:1077-1080

## ASSURE B Allele Specific Sequencing Using Restriction Enzyme and Biotinylation

Direct sequencing of PCR products to reveal mutations is only possible if the PCR product represents the target allele. If the PCR mixture is a heterogeneous mix of several alleles this method allows allele specific sequencing. Biotinylated PCR products are digested with a frequent cutter. The unaffected allele is captured and PCR-able because it has both primers sites and is captured on the magnetic beads.

Zhang D.-X. and Hetwitt G.M. (1996) An effective method for allele-specific sequencing using restriction enzyme and biotinylation (ASSURE B). *Molecular Ecology* 5:591-594.

## BAC Bacterial Artificial Chromosome

Cloning vector for large DNA fragments.

Shizuya, H., B. Birren, U.-J. Kim, V. Mancino, T. Slepak, Y. Tachiiri and M. Simon (1992) Cloning and stable maintenance of 300-kilobase-pair fragments of human DNA in *Escherichia coli* using an F-factor-based vector. *Proc. Natl. Acad. Sci. USA.* 89, 8794-8797.

## BIL Backcross Introgression Lines

## BSA Bulked Segregant Analysis

A rapid mapping strategy suitable for monogenic qualitative traits. When DNA of ten plants are bulked into one pool, all alleles must be present. Two bulked pools of segregants differing for one trait, will differ only at the locus harbouring that trait.

Michelmore R.W., Paran I and R.V. Kesseli (1991) Identification of markers linked to disease resistance genes by Bulk Segregant Analysis: A rapid method

to detect markers in specific genomic regions by using segregating populations. *Proc.Natl.Acad.Sci.* 88:9828-9832.

## CAPS Cleaved Amplified Polymorphic Sequences

PCR amplified DNA (STS-, EST- or SCAR-product) which is not yet polymorphic is digested with restriction endonucleases to reveal polymorphisms in restriction sites.

Akopyanz N., N.O. Bukanov, T.U. Westblom and D.E. Berg (1992) PCR-based RFLP analysis of DNA sequence diversity in the gastric pathogen *Helicobacter pylori*. *Nucleic Acid Res.* 20(23):6221-6225 [berg@borcim.wustl.edu](mailto:berg@borcim.wustl.edu)

Konieczny A, Ausubel FM (1993) A procedure for mapping *Arabidopsis* mutations using co-dominant ecotype-specific PCR-based markers. *Plant J.* 4:403-410.

## CFLP Cleavase Fragment Length Polymorphism

Technique for mutation detection in DNA stretches with unknown sequences. The method is an alternative to SSCP or DGGE. The principle relies on denaturation (melting) of DNA. The ssDNA will assume folded hairpin-like structures which are unique to the nucleotide sequence. The endonuclease Cleavase I specifically cleaves the junction between ss and ds regions. Fragments are 5' labelled, electrophoretically separated and visualized. Polymorphic fragments indicate a mutation; their length is indicative for the position of the mutated site.

Brow M.A.D. et al (1996) *Focus (Life technology)* 18(1):2-5

## CSGE Conformation-Sensitive Gel Electrophoresis

A method that requires no special equipment or preparation of the DNA. It detects conformational differences between heteroduplexes and homoduplexes, which are detected in differences in electrophoretic migration that are accentuated by the use of a mild denaturing agent.

Ganguly A, Rock MJ, Prockop DJ (1993) CSGE for rapid detection of single-base differences in double-stranded PCR products and DNA fragments: evidence for solvent-induced bends in DNA heteroduplexes. *Proc. Natl. Acad. Sci. USA* 90:10325-10329. Or see review: Ganguly and Prockop (1995) Detection of mismatched bases in double-stranded DNA by gel electrophoresis. *Electrophoresis* 16:1830-1835.

## DAF DNA Amplification Fingerprinting

refers to DNA amplification using a single random primer of 8-10 bases. DAF products are analysed on polyacrylamide gels and detected by silver staining.

Caetano-Anollés G., B.J. Bassam and P.M. Gresshoff (1991) REVIEW DNA amplification fingerprinting: A strategy for genome analysis. *Plant Molec. Biol. Reporter* 9(4):294-307. Caetano-Anollés, G., B.J. Bassam and P.M. Gresshoff (1991) Single-primer DNA amplification fingerprinting. *Bio/technology* 9:553-557

## DAMD Directed Amplification of Minisatellite-region DNA

## DDGE Denaturing Detergent Gradient Gel Electrophoresis

Method based on differential electrophoretic migration of single versus double-stranded DNA, which can easily be detected by commonly used ethidium bromide staining methods. However, it still requires special apparatus for generating detergent gradients and optimization of the experimental conditions.

Sheffield VC, Cox DR, Lerman LS, Myers RM (1989) Attachment of a 40-base-pair G + C-rich sequence (GC-clamp) to genomic DNA fragments by the polymerase chain reaction results in improved detection of single-base changes. *Proc. Natl Acad Sci USA* 86:232-236.

**2-DDGE** Two-Dimensional DNA Gel Electrophoresis.

HMW-DNA fragments restricted with a rare-cutter restriction enzyme are separated by PFGE. After staining and photography the lane is cut from the gel and incubated in a solution containing another restriction enzyme. Subsequently, the gel-strip and placed on top of a new gel for electrophoretic separation in the second dimension.

*DeSchenzo R.A. and Wise R.P. (1996) Variation in the ratio of physical to genetic distance in intervals adjacent to Mla locus on barley chromosome 1H. Mol Gen Genet 251:472-482.*

**DDRT-PCR** Differential Display Reverse Transcriptase-PCR

This technique enables the amplification of differentially expressed mRNA deriving from identical genotypes exposed to different treatments.

*Liang P, Pardee AB (1992) Differential display of eukaryotic messenger RNA by means of the polymerase chain reaction. Science 257 : 967-971*

**DFP** DNA finger printing

General name for multi-locus techniques used for identification of individuals or mapping.

**DGGE** Denaturing Gradient Gel Electrophoresis.

Resolves partially denatured double-stranded DNA in precisely defined conditions of temperature and denaturant concentration. Different alleles may denature to various extents under such conditions, and migrate differently on DGGE acrylamide gels.

*Myers R.M. et al. (1985) Nucleic Acid Res. 13:3111-3129; Myers R.M., T. Maniatis and L.S. Lerman (1987) Detection and localization of single base changes by DGGE. Methods Enzymol. 155:501-527.*

**DH** Doubled Haploids

A progeny of doubled haploids derived from a heterozygous or F1 individual can serve as a mapping population. Making a DH is faster than RIL; DHs have a better resolution than a F2 progeny (no heterozygosity); DHs can be maintained infinitely.

**DOL** Dye-labeled Oligonucleotide Ligation

Method for detection of SNP using ligation reaction. Template is mixed with a universal downstream primer and two alternate upstream primers. The SNP allows the correct hybridisation of one upstream primer, and prevents ligation of the alternate primer. In the presence of ligase the down- and upstream oligo ligate to a new molecule having the correct fluorescent labels. Specific signal detection is based on FRET.

*Chen X., P.-Y. Kwok (1997) Template-directed dye-terminator incorporation (TDI) assay: A homogeneous DNA diagnostic method based on fluorescence resonance energy transfer. Nucleic Acids Res. 25:2347-353.*

**DOP-PCR** Degenerate Oligonucleotide Primed-PCR

The technique uses 22-mer primers of partially degenerate sequence in a dual annealing temperature protocol and can therefore amplify DNA in a general and non-specific manner.

*Telenius H., Carter N.P., Bebb C.E., Nordenskjold M., Ponder B.A.J., Tunnacliffe A. (1992) Degenerate oligonucleotide-primed PCR: general amplification of target DNA by a single degenerate primer. Genomics 13:718-725.*

**EST** Expressed Sequence Tag

PCR based marker. Highly specific oligos (16-20-mers) are designed by using

sequence information of a cDNA. The locus represents a functional gene and is located in an actively transcribed region of the genome.

reference??

## FIGE Field Inversion Gel Electrophoresis

Electrophoresis method (comparable to PFGE) for the separation of high molecular weight DNA.

*Van Daelen, RA., Jonkers JJ, Zabel P (1989) Preparation of megabase-sized tomato DNA and separation of large restriction fragments by field inversion gel electrophoresis (FIGE). Plant Mol. Biol. 12, 341-342.*

## FRET Fluorescence Resonance Energy Transfer

Detection method using a combination of fluorescence labels. The first fluorescent molecule is used to absorb the energy, then the energy is transferred to the second fluorescent molecule. Detection of a light signal from the second dye is diagnostic for the two dyes being in a close proximity. This means that certain molecular reaction took place, which is diagnostic for the presence of a certain allelic state.

*Chen X., P.-Y. Kwok (1997) Template-directed dye-terminator incorporation (TDI) assay: A homogeneous DNA diagnostic method based on fluorescence resonance energy transfer. Nucleic Acids Res. 25:2347-353.*

## GMS Genomic Mismatch Scanning

## GS Genomic Selection

The number of polymorphic bands resembling the recurrent parent are used as the selection criterion.

*Lande R. and R. Thompson (1990) Efficiency of Marker-Assisted Selection in the Improvement of quantitative traits. Genetics 124:743-756.*

**HAPPY** An *in vitro* linkage technique based on screening approximately HA ploidy amounts of DNA by the PolYmerase chain reaction.

How HAPPY mapping of bacterial genomes works: Intact genomic DNA is broken by irradiation (I) to give a pool of random fragments, which are size selected by PFGE (II). A mapping panel of 96 aliquots is taken (III) from this pool. Each aliquot contains ~1 haploid genome's worth of DNA, so each marker is present in only a subset of the aliquots. (IV) The panel is pre-amplified a few hundred-fold by primer extension preamplification (PEP). (V) Subfractions of the pre-amplified panel are screened for specific markers using nested PCR. Markers A and B, (e.g.), are found to cosegregate frequently. This implies that they tend to lie on the same fragment of DNA, and so must be tightly linked. A map may be constructed (VI) by examining cosegregation frequencies between all markers. The number of polymorphic bands resembling the recurrent parent are used as the selection criterion.

*Piper MB, Bankier AT, and Paul H. Dear (1998) A HAPPY Map of *Cryptosporidium parvum*. Genome Res. 8(12):1299-1307.*

## IPCR Inverse Polymerase Chain Reaction

A technique to amplify genomic DNA flanking the insertion site of a transposon or T-DNA construct. The obtained flanking genomic DNA can be used as RFLP-probe to determine the map position of the insertion site of the construct.

*Triglia T., M.G. Peterson and D.J. Kemp (1988) A procedure for *in vitro* amplification of DNA segments that lie outside the boundaries of known sequences. Nucleic Acids Res. 16:8180-8186.*

## IRI Intermated Recombinant Inbreds

Just another structure of a mapping population like F2, BC, RIL, NIL. The

intermating of F2 individuals result in new recombination events. Therefore, IRI populations have improved genetic resolution.

Liu S.-C., S.P. Kowalski, T.-H. Lan, K.A. Feldmann and A.H. Paterson (1996) *Genome-wide high-resolution mapping by recurrent intermating using Arabidopsis thaliana as a model species. Genetics 142:247-258.*

## ISSR Inter-Simple Sequence Repeat amplification

PCR-based multi-locus marker system using oligonucleotide primers homologous to SSR sequences (such as (GATA)<sub>n</sub>). To avoid stutterings these primers can be anchored to unique genomic sequences flanking the repeat. Amplification products are only obtained in case SSRs in opposite orientation are found within a PCR-able distance, with a flanking sequences matching the oligo's. 3'-anchoring give better results than 5'-anchoring. Repeat polymorphisms within the SSR do not influence the chance for ISSR polymorphisms.

Kantety R.V., X. Zeng, J.L. Bennetzen and B. Zehr (1995) *Assessment of genetic diversity in dent and popcorn (Zea mays L.) inbred lines using Inter-Simple Sequence Repeat (ISSR) amplification. Molecular Breeding 1:365-373.*

## ISTR Inverse Sequence-Tagged Repeats

PCR-based multi-locus marker system using oligonucleotide primers homologous to dispersed high copy sequences, e.g. copia like transposons. Multi-locus polymorphic amplification products are obtained with two primers, designed in a direction outward the element, and allows the amplification of the stretch connecting the elements.

Rohde W. (1996) *Inverse sequence-tagged repeat (ISTR) analysis, a novel and universal PCR-based technique for genome analysis in the plant and animal kingdom. Journal of Genetics & Breeding 50(3): 249-261.*

## LCR Ligase Chain Reaction

Technique to detect a single-base substitution in a known sequence. The ligase enzyme links two adjacent oligo's which hybridize against the target sequence. In case of a mismatch the ligation of the adjacent oligo's will fail. Ligated oligo's resemble the target sequence and are available as target sequence for unligated oligo's in subsequent cycles resulting in an exponential amplification of ligated oligos.

Barany F. (1991) *Genetic disease detection and DNA amplification using cloned thermostable ligase. Proc. Natl. Acad. Sci. 88:189-193.*

## LOD Log Odds Ratio

A statistical measure indicating the significance of linkage. The  $10 \log$  of the Odds Ratio. The Odds Ratio is the probability (given  $H_0$ ) divided by the probability (given  $H_a$  = unlinked)

Risch N. (1992) *Genetic linkage: interpreting LOD scores. Science 255:803-804.*

## MAAP Multiple Arbitrary Amplicon Profilng

Basically similar to DAF or AFLP, but using multiple endonuclease digestions of template DNA or amplification products, significant increase of polymorphisms in fingerprints are generated.

Caetano-Anollés G., Bassam B.J. and Gresshoff P.M. (1993) *Enhanced detection of polymorphic DNA by multiple arbitrary amplicon profiling of endonuclease-digested DNA: identification of markers tightly linked to the supernodulation locus in soybean. Molec. Gen. Genet. 241:57-64.*

## MADGE Microtiter Array Diagonal Gel Electrophoresis

An easily scored difference in the electrophoretic migration of a PCR product can be obtained if an SNP alters the recognition sequence for a restriction endonuclease.

This principle has been used in the high through put, low-resolving microtiter array diagonal gel electrophoresis technique (MADGE). Here, restriction-digested PCR products are loaded on stackable horizontal gels with wells arranged in a microtiter format. The electric field is applied at an angle relative to the columns and rows of wells in the gel, allowing products from large numbers of reactions to be resolved between the downstream wells.

Day, I.N.M. and S.E. Humphries (1994) *Electrophoresis for genotyping: Microtiter array diagonal gel electrophoresis on horizontal polyacrylamide gels, hydrolink, or agarose*. *Anal. Biochem.* 222: 389B395.

## **MARS Marker Assisted Recurrent Selection**

A breeding strategy with the repeated application of indirect selection

## **MAS Marker Assisted Selection**

A breeding strategy applying indirect selection

## **MASDA Multiplexed Allele-Specific Diagnostic Assay**

Allele-specific oligo-nucleotide (ASO) hybridizations are frequently performed with either the DNA samples or the oligo-nucleotides arrayed on a surface, sometimes referred to as formats 1 and 2, respectively. Format 1 is suitable for screening many patient samples for the presence of a few disease-causing alleles, as in the multiplexed allele-specific diagnostic assay (MASDA), in which a pool of mutation-specific oligonucleotides are hybridized to spots of pooled PCR products from individual patients, and positive reactions are eluted and sequenced to determine which probe bound to a particular patient sample.

Shuber, A.P., L.A. Michalowsky, G.S. Nass, J. Skoletsky, L.M. Hire, S.K. Kotsopoulos, M.F. Phipps, D.M. Barberio, and K.W. Klinger (1997) *High throughput parallel analysis of hundreds of patient samples for more than 100 mutations in multiple disease genes*. *Hum. Mol. Genet.* 6: 337B347.

## **MATS Marker Addition Through Substraction**

A procedure based on subtractive hybridization and PCR amplification, for generating microsatellite-based markers directly from yeast artificial chromosomes (YACs). This strategy, termed MATS (marker addition through subtraction), exploits the fact that the only difference between a yeast host strain harboring a YAC and the host strain alone is the artificial chromosome. Given the low complexity of the yeast genome and relatively large target size presented by a YAC, only a single round of subtraction is required before amplification of the target sequences (YAC) and cloning into a plasmid vector for further analysis.

Chen-H; Pulido-JC; Duyk-GM (1995) *MATS: a rapid and efficient method for the development of microsatellite markers from YACs*. *Genomics*. 1995 Jan 1; 25(1): 1-8.

## **MB Molecular Beacons**

These oligonucleotide hybridization probes have two complementary DNA sequences flanking the target-specific sequence, and a donor-acceptor dye pair is present at opposite ends of each probe. When not hybridized to a target sequence, the probes adopt a hairpin-loop conformation, bringing the fluorophore and quencher pair close together, thereby extinguishing the donor fluorescence. On the other hand, when hybridized to the correct target sequence, the two dyes are separated and fluorescence increases by up to 900-fold. A valuable side effect of the preorganized hairpin probe design is that mismatch hybridization is further destabilized, providing increased allele selectivity in SNP analyses.

Tyagi, S. and F.R. Kramer. 1996. *Molecular beacons: Probes that fluoresce upon hybridization*. *Nature Biotechnol.* 14: 303B308.

Tyagi, S., D.P. Bratu, and F.R. Kramer. 1998. *Multicolor molecular beacons for allele discrimination*. *Nature Biotechnol.* 16: 49B53.

**MITE** Miniature Inverted-repeat Transposable Element

A class of transposable elements, which share features of both class I and class II elements, and, therefor, remain unclassified. The transposons in this group share structural, but not sequence similarity.

Wessler S, Bureau TE, White SE (1995) LTR-retrotransposons and MITEs: important players in the evolution of plant genomes. *Curr. Op. Genet. Dev.* 5:814-821.

**MSAP** Methylation Sensitive Amplification Polymorphism

A class of transposable elements, which share features of both class I and class II elements, and, therefor, remain unclassified. The transposons in this group share structural, but not sequence similarity.

Reyna-Lopez GE, Simpson J, Ruiz-Herrera J (1997) Differences in DNA methylation patterns are detectable during the dimorphic transition of fungi by amplification of restriction polymorphisms. *Mol Gen Genet* 253:703-710.

Xiong LZ, Xu CG, Saghai Maroof MA, Qifa Zhang (1999) Patterns of cytosine methylation in an elite rice hybrid and its parental lines, detected by a methylation-sensitive amplification polymorphism technique. *Mol Gen Genet* 261:439-446.

**NASBA** Nucleic Acid Sequence Based Amplification

This method has the unique ability to amplify a specific RNA sequence in the presence of homologous DNA sequences, under iso-thermal (=no PCR) conditions. NASBA employs reverse transcriptase and primers to generate a double stranded cDNA template with a T7 promoter. T7 RNA polymerase then initiates transcription and generates multiple copies of RNA, which re-enters the cycle to generate cDNA.

Sooknanan R., and L.T. Malek (1995) NASBA a detection and amplification system uniquely suited for RNA. *Bio/Technology* 13:563-564.

**NIL** Near Isogenic Lines

Homozygous line, produced by recurrent selection, which only heterozygous and different from the recurrent parent with respect to a locus of interest.

Young N.D., D. Zamir, M.W. Ganal and S.D. Tanksley (1988) Use of isogenic lines and simultaneous probing to identify DNA markers tightly linked to the *Tm-2a* gene in tomato. *Genetics* 120:579-585.

**OLA** Oligonucleotide Ligation Assay

In the OLA, the sequence surrounding the SNP is first amplified by PCR, whereas in LCR, genomic DNA can be used as a template. By use of allele-specific ligation probes differentially labeled with lanthanide chelates, and a multipronged solid support for parallel sample handling, sets of ligation reactions can be conveniently analyzed via time-resolved fluorometry (Samiotaki et al. 1994). Dual-color detection of allele-specific ligation products is also possible in a regular spectrophotometer (Tobe et al. 1996).

Samiotaki, M., M. Kwiatkowski, J. Parik, and U. Landegren (1994) Dual-color detection of DNA sequence variants by ligase-mediated analysis. *Genomics* 20: 238B242.

Tobe, V.O., S.L. Taylor, and D.A. Nickerson (1996) Single-well genotyping of diallelic sequence variations by a two-color ELISA-based oligonucleotide ligation assay. *Nucleic Acids Res.* 24: 3728B3732.

**OP** Oligonucleotide Polymorphism

PCR based markers using two 18-mer oligonucleotide primers used for tagging specific loci or alleles (see ASO). Primer design is based on known cDNA sequences.

Beckmann J.S. (1988) REVIEW Oligonucleotide polymorphisms: A new tool for



*genomic genetics. Bio/Technology 6:161-164.*

## PCR Polymerase Chain Reaction

The exponential amplification of DNA fragment(s) using oligonucleotide(s), a thermostable DNA polymerase and repeated cycles of DNA denaturing, primer-annealing and primer-extension.

*Mullis K.B. and F.A. Faloona (1987) Specific synthesis of DNA in vitro via the polymerase catalyzed reaction. Meth. Enzymol. 255:335-350. Erlich H.A., D. Gelfand and J.J. Sninsky (1991) Recent advances in the polymerase chain reaction. Science 252:1643-1651*

## PFGE Pulsed Field Gel Electrophoresis

Gel system to separate DNA fragments sizing .5 to 3 Megabases with alternating electric fields.

*Schwartz D.C. and C.R. Cantor (1984) Cell 37:67-75*

## PIC Polymorphic Information Content

A measure for the probability of polymorphism at a (STMS-) locus, calculated from the number of alleles and their respective allele frequencies in a population.

*Weber J.L. (1990) Informativeness of human (dC-dA)<sub>n</sub>-(dG-dT)<sub>n</sub> polymorphisms. Genomics 7:524-530.*

## PRINS Primed in situ labelling

beschrijving

referentie

## QTL Quantitative Trait Locus

Single locus from a series of polygenes which are involved in a quantitative trait.

*Paterson A.H., E.S. Lander, J.D. Hewitt, S. Peterson, S.E. Lincoln, S.D. Tanksley (1988) Resolution of quantitative traits into Mendelian factors using a complete linkage map of RFLPs. Nature 334:721-726.*

## RAMCM RNase A Mismatch Cleavage Method

For detecting single base substitutions in ma:ma or ma:dna hybrids with ribonuclease A.

*Lopez-Galindez et al (1995) PNAS 88:4280-84; Myers et al (1985) Science 230:1242-46*

## RAMP Random Amplified Multiple Polymorphisms ??look up??

*Zietkiewicz E., A. Rafalski and D. Labuda (1994) Genome fingerprinting by simple sequence repeats (SSR)-anchored PCR amplification. Genomics 20:176-183.*

## RAPD Random Amplified Polymorphic DNA

A PCR product that is obtained from genomic DNA using a single or a combination of typically 10-mer oligonucleotides. Alleles are visualized by the fragments that are amplified, separated on agarose gels and stained with EtBr. RAPDs show dominant inheritance. Variation is based on the position and orientation of primer-annealing sites and the interval they span.

*Williams J.G.K., A.R. Kubelik, K.J. Livak, J.A. Rafalski and S.V. Tingey (1990) DNA polymorphisms amplified by arbitrary primers are useful as genetic*

markers. *Nucleic Acid Research* 18:6531-6535.

## **RDA** Representational Display Analysis

Lisitsyn.... something on genomic subtraction??

## **REP-PCR** Repat sequence primed PCR

## **REMI** Restriction Enzyme Mediated Integration

Method to stimulate transformation. The enzyme enters the cell during electroporation and facilitates intergration of linearized foreign DNA with compatible ends into the host chromosomes.

*Kuspa A. and W.F. Loomis (1992) Tagging developmental genes in Dictyostelium by restriction enzyme-mediated integration of plasmid DNA. Proc. Natl. Acad. Sci. USA 89:8803-8807*

## **REMI-RFLP** Restriction Enzyme Mediated Integration - Restr. Fragm. Length Polym.

Length differences caused by REMI are used as a polymorphic marker locus.

*Kuspa A. and W.F. Loomis (1994) REMI-RFLP mapping in the Dictyostelium genome. Genetics 138:665-674.*

## **RICH** Rapid Isolation of c-DNA by Hybridization

Method depends on solution hybridization, enzymatic modification and amplification/ selection of sequences present in both cDNA populations and the genomic clones.

*Hamaguchi, M., E.A. O'Connor, T. Chen, L. Parnell, R.W. McCombie, and M.H. Wiggler (1998) Rapid isolation of cDNA by hybridization. Proc. Natl. Acad. Sci. USA 95(7):3764-3769.*

## **RIL** Recombinant Inbred Lines

An F4 population derived from a F2 by single seed descent (SSD). The consequence of infinite selfing is a doubling of the recombination frequency; a immortalization of the population and the exclusion of dominance variance due to heterozygosity.

*Burr B. and F.A. Burr (1991) REVIEW Recombinant inbreds for molecular mapping in maize. Trends in Genetics 7:55-60*

## **RBIP** Retrotransposon-Based Insertion Polymorphisms

*Flavell AJ, M.R. Knox, S.R. Pearce and T.H.N. Ellis (1998) Retrotransposon-based insertion polymorphisms (RBIP) for high throughput marker analysis. Plant J 16(5):*

## **RCA** Rolling Circle Amplification

A DNA amplification method (alternatives to PCR or NASBA). A probe is hybridised to the target sequence to form an open circle, because the ends are inversely complementary to the target sequence. With ligase the circular probe can be closed and will surround the target sequence as a padlock. A single primer homologous to the non-complementary part of the circle is extended by phage  $\phi$  29 DNA polymerase. Copies of the target sequence roll from the circle in a long sequence of tandem repeats.

*Lizardi PM, X Huang, Z Zhu, P Bray-Ward, DC Thomas & DC Ward (1998) Mutation detection and single-molecule counting using isothermal rolling-circle*

amplification. *Nature Genetics* 19:225-232 (US Patent 5 854 033 ,29 December 1988).

## **RFLP** Restriction Fragment Length Polymorphism

A DNA fragment used to probe Southern blots of restricted genomic DNA from different strains of the same species. This results in the visualisation of variation in the size and/or number of detected restriction fragments generated from the different strains. The detected length variation is based on DNA sequence variation caused by insertions, deletions or changes in restriction sites.

Botstein D., R.L. White, M. Skolnick and R.W. Davis (1980) Construction of a genetic linkage map in man using restriction fragment length polymorphisms. *Am. J. Hum. Genet.* 32:314-331.

Soller M. and Beckmann J.S. (1983) Genetic polymorphism in varietal identification and genetic improvements. *Theor. Appl. Genet.* 67:25-33

## **SAMPL** Selective Amplification of Microsatellite Polymorphic Loci

Based on AFLP. The template is identical to AFLP template, but the rare cutter primer is replaced by a (CA)<sub>72</sub>(TA)<sub>22</sub> or (GT)<sub>72</sub>(AT)<sub>2</sub>.

Dean L, Witsenboer H, Kesseli R, Paran I. and R.W. Michelmore (1996) Identification of makers clusters of disease resistance genes in lettuce. *Plant Genome IV Poster abstract.*

Morgante M and Vogel J: Compound microsatellite primers for the detection of genetic polymorphisms. U.S. Patent Appl 08/326456 (1994).

Paglia G., M. Morgante (1998) PCR-based multiplex DNA fingerprinting techniques for the analysis of conifer genomes. *Molecular Breeding* 4:173-177.

## **SCAR** Sequence Characterized Amplified Regions

PCR based marker. A genomic DNA fragment at a single genetically defined locus that is identified by PCR amplification using a pair of specific (16-24-mer) oligonucleotide primers.

Paran I. and R.W. Michelmore (1993) Development of reliable PCR-based markers linked to downy mildew resistance genes in lettuce. *Theor. Appl. Genet* 85:985-993

## **SDA** Strand Displacement Amplification

DNA is amplified by a DNA-polymerase lacking exo-nuclease activity. Therefore, the newly synthesized strand displaces the former strand. The DNA-polymerase can start again from a single-strand nick generated by a restriction enzyme. The other strand is protected from the restriction enzyme by modified nucleotides incorporated during DNA synthesis.

Walker G.T., M.C. Little, J.G. Nadeau and D.D. Shank (1992) Isothermal in vitro amplification of DNA by a restriction enzyme/DNA polymerase system. *Proc. Natl. Acad. Sci.* 89:392-396.

## **SNAP** Single Nucleotide Amplified Polymorphisms

PCR based marker..

Mindrinus Ausubel

## **SNP** Single Nucleotide Polymorphisms

Polymorphism based on a nucleotide substitution. Used as a marker diagnostic for a specific trait. Often mentioned in connection with a technique which allows the specific recognition of the SNP.

Jordan S.A., P. Humphries (1994) Single nucleotide polymorphism in exon 2 of the BCP gene on 7q31-q35. *Human Molecular Genetics* 3(10): 1915

### **SPAR** Single Primer Amplification Reactions

PCR based marker. SSRs like (GATA)<sub>4</sub> are used to prime PCR on genomic DNA. The stretches of unique DNA in between the SSR are reproducibly amplified. Tetra-nucleotide repeats perform better than di- or tri-nucleotide repeats.

Gupta M., Y.-S. Chyi, J. Romero-Severson and J.L. Owen (1994) Amplification of DNA markers from evolutionarily diverse genomes using single primers of simple-sequence repeats. *Theor. Appl. Genet* 89:998-1006.

### **SRFA** Selective Restriction Fragment Amplification

A procedure taking oligo's that are elongated with selective bases. Because of these bases not all restriction fragments can be amplified in a PCR reaction.

Zabeau M. (1992) Gast-College hybride rassen, Vakgroep Plantenveredeling LUW.

### **S-SAP** Sequence Specific Amplification Polymorphisms

DNA template is prepared analogous to AFLP. Fingerprints are generated using a radioactively labelled primer homologous to the Long Terminal Repeat. The other primer fits to the template anchor sequence. By adding selective nucleotides to the anchored primer, subsets of amplification products are generated in a PCR reaction.

Waugh R, et al. (1997) Genetic distribution of Bare-1-like retrotransposable elements in the barley genome revealed by sequence-specific amplification polymorphism (S-SAP).

### **SSCP** Single Stranded Confirmational Polymorphism

Relies on secondary and tertiary structure differences between denatured and rapidly cooled amplified DNA fragments that differ slightly in their DNA sequence. Different SSCP alleles are resolved on non-denaturing acrylamide gels, usually at low temperatures. The ability to resolve alleles depends on the conditions of electrophoresis.

Orita M., Y. Suzuki, T. Sekiya and K. Hayashi (1989) Rapid and sensitive detection of point mutations and DNA polymorphisms using polymerase chain reaction. *Genomics* 5:874-879 (see also Hayashi (1991) *PCR Methods Appl* 1:34-38 or Antolin et al. (1996) *Genetics* 143:1727-1738.

### **SSI** Site-selected Insertion - PCR

SSI is a PCR-based mutagenesis and tagging technique which uses primers located within the transposon and a target gene for detection of transposon insertions into cloned genes.

Ballinger D.G. and S. Benzer (1989) Targeted gene mutations in *Drosophila*. *Proc. Natl. Acad. Sci.* 86:9402-9406

Koes R., et al. (1995) Targeted gene inactivation in petunia by PCR-based selection of transposon insertion mutants. *Proc. Natl. Acad. Sci.* 92:8149-8153

### **SSLP** Simple Sequence Length Polymorphism

Identical to SSR, The jargon is used in (for example) mouse genetics.

W Dietrich, H Katz, SE Lincoln, HS Shin, J Friedman, NC Dracopoli and ES Lander (1992) A genetic map of the mouse suitable for typing intraspecific crosses. *Genetics* 131:423-447.  
[http://www-genome.wi.mit.edu/genome\\_data/mouse/](http://www-genome.wi.mit.edu/genome_data/mouse/)

Bell CJ, Ecker JR (1994) Assignment of 30 microsatellite loci to the linkage map

of *Arabidopsis*. *Genomics* 19:137-144.

## SSR Simple Sequence Repeats

Synonymous to STR or micro satellite repeats, in particular the dinucleotide repeats  $(AC)_n$   $(AG)_n$   $(AT)_n$

*Akkaya M.S., A.A. Bhagwat and P.B. Cregan (1992) Length polymorphisms of simple sequence repeat DNA in soybean. Genetics 132:1131-1139;*

*Morgante M. and A.M. Olivieri (1993) The plant J. 3:175-182*

## SSR-anchored PCR See ISSR

## STMS Sequence Tagged Microsatellite Site

A PCR-based marker. The DNA sequence of unique DNA flanking microsatellites  $(GATA)_n$ ,  $(CT)_n$  etc. is used to construct 20-mer oligos. Amplification products spanning the microsatellite show rich allele divergency.

*Beckmann J.S. and M. Soller (1990) REVIEW Toward a unified approach to genetic mapping of eukaryotes based on sequence tagged microsatellite sites. BioTechnology 8:930-932.*

## STR Short Tandem Repeats

Synonymous to SSR detected as  $(TG)_n$ ,  $(CA)_n$

*Hamada H., M.G. Petrino and T. Kakunaga (1982) A novel repeat element with Z-DNA-forming potential is widely found in evolutionarily diverse eukaryotic genomes. Proc. Natl. Acad. Sci. USA 79:6465-6469*

*Hamada H., M.G. Petrino, T. Kakunaga, M. Seidman and B.D. Stollar (1984) Characterization of genomic poly(dT-dG)poly(dC-dA) sequences: Structure, organization and conformation. Mol. Cell. Biol. 4:2610-2621*

## STS Sequence Tagged Site

A PCR-based marker derived from a RFLP-probe, by constructing primers according to the end-sequence of the RFLP-probe.

*Olsen M., L. Hood, C. Cantor and D. Botstein (1989) A common language for physical mapping of the human genome. Science 245:1434-1435.*

## TaqMan

The TaqMan assay takes advantage of the 5'-nuclease activity of Taq DNA polymerase to digest a DNA probe annealed specifically to the accumulating amplification product (Holland et al. 1991). TaqMan probes are labeled with a donor-acceptor dye pair that interact via fluorescence resonance energy transfer (FRET). Cleavage of the TaqMan probe dissociates the donor dye from the quenching acceptor dye, greatly increasing the donor fluorescence. All reagents necessary to detect two allelic variants can be assembled at the beginning of the reaction and the results are monitored in real time during PCR.

*Livak, K., J. Marmaro, and J.A. Todd. 1995. Towards fully automated genome-wide polymorphism screening. Nature Genet. 9: 341B342.*

## TGGE Temperature-Gradient Gel Electrophoresis

Method based on differential electrophoretic migration of single versus double-stranded DNA, which can easily be detected by commonly used ethidium bromide staining methods. However, it still requires special apparatus for generating temperature gradients and optimization of the experimental conditions.

*Wartell RM, Hosseini SH, Moran CP (1990) Detecting base pair substitutions in*

*DNA fragments by temperature-gradient gel electrophoresis. Nucleic Acids Res. 18:2699-2705.*

## VNTR Variable Number Tandem Repeat

A genetic locus studied from a Southern blot, probed with a labelled minisatellite repeat. It's used in fingerprinting and forensic studies. It is not similar to a STMS. The core units vary in lenght from 11 to 60 bp.

*Nakamura Y., M. Leppert, P. O'Connell, R. Wolff, T. Holm, M. Culver, C. Martin, E. Fujimoto, M. Hoff, E. Kumlin and R. White (1987) Variable number tandem repeat (VNTR) markers for human gene mapping. Science 235:1616-1622.*

*Jeffreys A.J., V. Wilson and S.L. Thein (1985) Hypervariable "minisatellite" regions in human DNA. Nature 314:67-73.*

## YAC Yeast Artificial Chromosome

A plasmid that contains all the sequences necessary for stable maintenance in yeast (a centromere, DNA replication origin and telomeres), as well as a yeast selectable marker.

*Schlessinger D. (1990) REVIEW Yeast artificial chromosomes: tools for mapping and analysis of complex genomes. Trends in Genetics 6:248-258.*

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